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Kommentar zu: E. Frey, St. Anna Kinderspital, Wien – Zweitumoren nach Krebserkrankungen im Kindes- und Jugendalter.

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- Dörr W, Herrmann Th (2002) Second primary tumors after radiotherapy for malignancies. Treatment related parameters. *Strahlenther Onkol* 178:357–362
- Frey E (2010) Zweitumoren nach Krebserkrankungen im Kindes- und Jugendalter. *Forum* 25:39–41
- Trott KR (2009) Second cancers after radiotherapy. In: Joiner M, Van der Kogel A (eds) *Basic clinical radiobiology*, 4th edn, chap 25. Hodder Arnold, London, pp 339–352

— Literatur Seite 15-19

Möglichkeiten und Gefahren der prädiktiven Diagnostik. Plädoyer für die stärkere Berücksichtigung des Themas „soziale Ungleichheit und Gesundheit“

- Lampert T, Saß A-C, Häfeling M, Ziese T (2005) Armut, soziale Ungleichheit und Gesundheit. Gesundheitsberichterstattung des Bundes: Expertise des Robert Koch-Instituts zum 2. Armuts- und Reichtumsbericht der Bundesregierung
- Mielck A, Rogowski W (2007) Bedeutung der Genetik beim Thema „soziale Ungleichheit und Gesundheit“. *Bundesgesundheitsbl* 50:181–191
- Mackenbach J (2005) Genetics and health inequalities: hypotheses and controversies. *J Epidemiol Community Health* 59:268–273
- Rauprich O (2009) Gesundheitsgerechtigkeit. Zur Theorie von Norman Daniels. *Bundesgesundheitsbl* 52:519–526
- Sachverständigenrat zur Begutachtung der Entwicklung im Gesundheitswesen. Sondergutachten 2009: Koordination und Integration – Gesundheitsversorgung in einer Gesellschaft des längeren Lebens (Kurzfassung)
- Berth H, Dinkel A, Balck F (2004) Das Vertrauen der deutschen Bevölkerung in die Durchführung und Ergebnisverwendung genetischer Untersuchungen. Ergebnisse einer Repräsentativstudie. *J Public Health* 12:105–110
- Rogowski WH, Grosse SD, Khoury MJ (2009) Challenges of translating genetic tests into clinical and public health practice. *Nat Rev Genet* 10(7):489–495
- Schleberger B, Fonatsch C (2007) Einführung zum Schwerpunktthema „Familiäre Krebserkrankungen“. *Med Genet* 19(2):189–190
- Rees R, Apps P (2006) Genetic testing, income distribution and insurance markets. *Ann Econ Statist* 83/84:353–368

— Literatur Seite 23-25

Genetische Diagnostik erblicher Tumorerkrankungen. Wie zuverlässig ist die Diagnostik?

- Aretz S (2010) Differentialdiagnostik und Früherkennung hereditärer gastrointestinaler Polyposis-Syndrome. *Dtsch Arztebl* 107:163–173
- Aretz S, Propping P, Nöthen MM (2006) Indikationen zur molekulargenetischen Diagnostik bei erblichen Krankheiten. Zertifizierte medizinische Fortbildung. *Dtsch Arztebl* 103:A550–A560
- Engel C, Forberg J, Holinski-Feder E et al (2006) Novel strategy for optimal sequential application of clinical criteria, immunohistochemistry and microsatellite analysis in the diagnosis of hereditary nonpolyposis colorectal cancer. *Int J Cancer* 118:115–122
- Umar A, Boland CR, Terdiman JP et al (2004) Revised Bethesda Guidelines for hereditary nonpolyposis colorectal cancer (Lynch syndrome) and microsatellite instability. *J Natl Cancer Inst* 96:261–268
- Vasen HF, Watson P, Mecklin JP, Lynch HT (1999) New clinical criteria for hereditary nonpolyposis colorectal cancer (HNPCC, Lynch syndrome) proposed by the international collaborative group on HNPCC. *Gastroenterology* 116(6):1453–1456

— Literatur Seite 26-30

Erbliche Formen des Darmkrebs. Häufigkeit, Diagnostik, klinische Aspekte und Besonderheiten im Management

- Hampel H, Frankel WL, Martin E et al (2005) Screening for the Lynch syndrome (hereditary nonpolyposis colorectal cancer). *N Engl J Med* 352(18):1851–1860
- Vasen HF, Watson P, Mecklin JP, Lynch HT (1999) New clinical criteria for hereditary nonpolyposis colorectal cancer (HNPCC, Lynch syndrome) proposed by the International Collaborative Group on HNPCC. *Gastroenterology* 116(6):1453–1456

- Lynch HT, Boland CR, Gong G et al (2006) Phenotypic and genotypic heterogeneity in the Lynch syndrome: diagnostic, surveillance and management implications. *Eur J Hum Genet* 14(4):390–402
- Vasen HF, Mecklin JP, Khan PM, Lynch HT (1991) The International Collaborative Group on Hereditary Non-Polyposis Colorectal Cancer (ICG-HNPCC). *Dis Colon Rectum* 34(5):424–425
- Umar A, Boland CR, Terdiman JP et al (2004) Revised Bethesda Guidelines for hereditary nonpolyposis colorectal cancer (Lynch syndrome) and microsatellite instability. *J Natl Cancer Inst* 96(4):261–268
- Hampel H, Stephens JA, Pukkala E et al (2005) Cancer risk in hereditary nonpolyposis colorectal cancer syndrome: later age of onset. *Gastroenterology* 129(2):415–421
- Watson P, Vasen HF, Mecklin JP et al (2008) The risk of extra-colonic, extra-endometrial cancer in the Lynch syndrome. *Int J Cancer* 123(2):444–449
- Ten Kate GL, Kleibeuker JH, Nagengast FM et al (2007) Is surveillance of the small bowel indicated for Lynch syndrome families? *Gut* 56(9):1198–1201
- Schulmann K, Engel C, Propping P, Schmiegel W (2008) Small bowel cancer risk in Lynch syndrome. *Gut* 57(11):1629–1630
- Goecke T, Schulmann K, Engel C et al (2006) Genotype-phenotype comparison of German MLH1 and MSH2 mutation carriers clinically affected with Lynch syndrome: a report by the German HNPCC Consortium. *J Clin Oncol* 24(26):4285–4292
- Plaschke J, Engel C, Kruger S et al (2004) Lower incidence of colorectal cancer and later age of disease onset in 27 families with pathogenic MSH6 germline mutations compared with families with MLH1 or MSH2 mutations: the German Hereditary Nonpolyposis Colorectal Cancer Consortium. *J Clin Oncol* 22(22):4486–4494
- Järvinen HJ, Aarnio M, Mustonen H et al (2000) Controlled 15-year trial on screening for colorectal cancer in families with hereditary nonpolyposis colorectal cancer. *Gastroenterology* 118(5):829–834
- Järvinen HJ, Renkonen-Sinisalo L, Aktán-Collán K et al (2009) Ten years after mutation testing for Lynch syndrome: cancer incidence and outcome in mutation-positive and mutation-negative family members. *J Clin Oncol* 27(28):4793–4797
- Vasen HF, Abdurahman M, Brohet R et al (2010) One to 2-year surveillance intervals reduce risk of colorectal cancer in families with lynch syndrome. *Gastroenterology* [Epub ahead of print]
- Jong AE de, Hendriks YM, Kleibeuker JH et al (2006) Decrease in mortality in Lynch syndrome families because of surveillance. *Gastroenterology* 130(3):665–671
- Engel C, Rahner N, Schulmann K et al (2010) Efficacy of annual colonoscopic surveillance in individuals with hereditary nonpolyposis colorectal cancer. *Clin Gastroenterol Hepatol* 8(2):174–182
- Schmiegel W, Pox C, Adler G et al (2004) Deutsche Gesellschaft für Verdauungs- und Stoffwechselkrankheiten. S3-Leitlinien-Konferenz „Kollektives Karzinom“ 2004. *Z Gastroenterol* 42(10):1129–1177
- Dove-Edwin I et al (2002) The outcome of endometrial carcinoma surveillance by ultrasound scan in women at risk of hereditary nonpolyposis colorectal carcinoma and familial colorectal carcinoma. *Cancer* 94:1708–1712
- Rijcken FE, Mourits MJ, Kleibeuker JH et al (2003) Gynecologic screening in hereditary nonpolyposis colorectal cancer. *Gynecol Oncol* 91:74–80
- Vasen HF et al (2007) Guidelines for the clinical management of Lynch syndrome (hereditary non-polyposis cancer). *J Med Genet* 44:353–362
- Dijkhuizen FP, Mol BW, Brolmann HA, Heintz AP (2000) The accuracy of endometrial sampling in the diagnosis of patients with endometrial carcinoma and hyperplasia: a meta-analysis. *Cancer* 89:1765–1772
- Patai K, Szentmariay IF, Jakab Z, Szilagy G (2002) Early detection of endometrial cancer by combined use of vaginal ultrasound and endometrial vacuum sampling. *Int J Gynecol Cancer* 12:261–264
- Madari S, Al-Shabibi N, Papalampros P et al (2009) A randomised trial comparing the H Pipelle with the standard Pipelle for endometrial sampling at 'no-touch' (vaginoscopic) hysteroscopy. *BJOG* 116:32–37
- Braun MS, Richman SD, Quirke P et al (2008) Predictive biomarkers of chemotherapy efficacy in colorectal cancer: results from the UK MRC FOCUS trial. *J Clin Oncol* 26(16):2690–2698
- Sargent DJ, Marsoni S, Thibodeau SN et al (2008) Confirmation of deficient mismatch repair (dMMR) as a predictive marker for lack of benefit from 5-FU based chemotherapy in stage II and III colon cancer (CC): a pooled molecular reanalysis of randomized chemotherapy trials. *J Clin Oncol* 20 (Suppl): abstr 4008
- Tejpar S, Bosman F, Delorenzi M et al (2009) Microsatellite instability (MSI) in stage II and III colon cancer treated with 5FU-LV or 5FU-LV and irinotecan (PETACC 3-EORTC 40993-SAKK 60/00 trial). *J Clin Oncol* 27 (Suppl):15 s, abstr 4001
- Kim ST, Lee J, Park SH et al (2009) Clinical impact of microsatellite instability in colon cancer following adjuvant FOLFOX therapy. *Cancer Chemother Pharmacol* [Epub ahead of print]
- Schmeler KM, Lynch HT, Chen LM et al (2006) Prophylactic surgery to reduce the risk of gynecologic cancers in the Lynch syndrome. *N Engl J Med* 354(3):261–269

29. Spigelman AD, Williams CB, Talbot IC et al (1989) Upper gastrointestinal cancer in patients with familial adenomatous polyposis. *Lancet* 2(8666):783–785
30. Groves CJ, Saunders BP, Spigelman AD, Phillips RK (2002) Duodenal cancer in patients with familial adenomatous polyposis (FAP): results of a 10 year prospective study. *Gut* 50:636–641
31. Saurin JC, Gutknecht C, Napoleon B et al (2004) Surveillance of duodenal adenomas in familial adenomatous polyposis reveals high cumulative risk of advanced disease. *J Clin Oncol* 22:493–498
32. Burke CA, Santisi J, Church J, Levinthal G (2005) The utility of capsule endoscopy small bowel surveillance in patients with polyposis. *Am J Gastroenterol* 100(7):1498–1502
33. Schulmann K, Hollerbach S, Kraus K et al (2005) Feasibility and diagnostic utility of video capsule endoscopy for the detection of small bowel polyps in patients with hereditary polyposis syndromes. *Am J Gastroenterol* 100(1):27–37
34. Galle TS, Juel K, Bulow S (1999) Causes of death in familial adenomatous polyposis. *Scand J Gastroenterol* 34:808–812
35. Burt RW, Leppert MF, Slattery ML et al (2004) Genetic testing and phenotype in a large kindred with attenuated familial adenomatous polyposis. *Gastroenterology* 127(2):444–451
36. Knudsen AL, Bisgaard ML, Bulow S (2003) Attenuated familial adenomatous polyposis (AFAP). A review of the literature. *Fam Cancer* 2(1):43–55
37. Sieber OM, Lipton L, Crabtree M et al (2003) Multiple colorectal adenomas, classic adenomatous polyposis, and germ-line mutations in MYH. *N Engl J Med* 348:791–799
38. Vogt S, Jones N, Christian D et al (2009) Expanded extracolonic tumor spectrum in MUTYH-associated polyposis. *Gastroenterology* 137(6):1976–1985
39. Jo WS, Bandipalliam P, Shannon KM et al (2005) Correlation of polyp number and family history of colon cancer with germline MYH mutations. *Clin Gastroenterol Hepatol* 3(10):1022–1028
40. Wang L, Baudhuin LM, Boardman LA et al (2004) MYH mutations in patients with attenuated and classic polyposis and with young-onset colorectal cancer without polyps. *Gastroenterology* 127(1):9–16. Erratum in: *Gastroenterology* 127(5):1651
41. Giardiello FM, Trimbath JD (2006) Peutz-Jeghers syndrome and management recommendations. *Clin Gastroenterol Hepatol* 4(4):408–415
42. Hearle N, Schumacher V, Menko FH et al (2006) Frequency and spectrum of cancers in the Peutz-Jeghers syndrom. *Clin Cancer Res* 12(10):3209–3215
43. Jass JR, Williams CB, Bussey HJ, Morson BC (1988) Juvenile polyposis – a precancerous condition. *Histopathology* 13(6):619–630
44. Howe JR, Mitros FA, Summers RW (1998) The risk of gastrointestinal carcinoma in familial juvenile polyposis. *Ann Surg Oncol* 5(8):751–756
13. Rolland JS, Williams J (2005b) Toward a biopsychosocial model for 21st-century genetics. *Fam Process* 44:3–24
14. Rolland JS (2005a) Cancer and the family: an integrative model. *Cancer* 104 (Suppl 11):2584–2595
15. Schmutzler R, Schlegelberger B, Meindl A et al (2003) Beratung, genetische Testung und Prävention von Frauen mit einer familiären Belastung für das Mamma- und Ovarialkarzinom. Interdisziplinäre Empfehlungen des Verbundprojekts „Familiärer Brust- und Eierstockkrebs“ der Deutschen Krebshilfe. *medgen* 15:385–395
16. Oostrom I van, Meijers-Heijboer H, Duivenvoorden HJ et al (2006) Experience of parental cancer in childhood is a risk factor for psychological distress during genetic cancer susceptibility testing. *Ann Oncol* 17:1090–1095

— Literatur Seite 34-37

Psychosoziale und familiäre Aspekte bei der genetischen Diagnostik von erblichem Darmkrebs. Heidelberger Interdisziplinäre Sprechstunde für Familiären Darmkrebs

1. Bish A, Sutton S, Jacobs C et al (2002) No news is (not necessarily) good news: impact of preliminary results for BRCA1 mutation searches. *Genet Med* 4:353–358
2. Bonadona V, Saltel P, Desseigne F et al (2002) Cancer patients who experienced diagnostic genetic testing for cancer susceptibility: reactions and behavior after the disclosure of a positive test result. *Cancer Epidemiol Biomarkers Prev* 11:97–104
3. Carlsson C, Nilbert M (2007) Living with hereditary non-polyposis colorectal cancer: experiences from and impact of genetic testing. *J Genet Couns* 16:811–820
4. Claes E, Evers-Kiebooms G, Boogaerts A et al (2004) Diagnostic genetic testing for hereditary breast and ovarian cancer in cancer patients: women's looking back on the pre-test period and a psychological evaluation. *Genet Test* 8:13–21
5. Domanska K, Nilbert M, Soller M et al (2007) Discrepancies between estimated and perceived risk of cancer among individuals with hereditary nonpolyposis colorectal cancer. *Genet Test* 11:183–186
6. Gritz E, Peterson S, Vernon SW et al (2005) Psychological impact of genetic testing for hereditary nonpolyposis colorectal cancer. *J Clin Oncol* 23:1902–1910
7. Jost R, Schroeter C, Kloor M et al (2010) Disclosure of MSI results to high risk families for HNPCC: how well are they understood? (submitted)
8. Keller M, Jost R et al (2002) Comprehensive genetic counselling for families at risk for HNPCC: impact on distress and perceptions. *Genet Test* 6:291–301
9. Keller M, Jost R, Haunstetter CM et al (2008) Psychosocial outcome following genetic risk counselling for familial colorectal cancer. A comparison of affected patients and family members. *Clin Genet* 74:414–424
10. Keller M (2010) Erbliche Tumorerkrankungen – psychosoziale und familiäre Aspekte. Psychotherapie im Dialog (in press)
11. McDaniel SH (2005) The psychotherapy of genetics. *Fam Process* 44:25–44
12. Meiser B (2005) Psychological impact of genetic testing for cancer susceptibility: an update of the literature. *Psychooncology* 14:1060–1074